



## MCCC1 gene

methylcrotonoyl-CoA carboxylase 1

### Normal Function

The *MCCC1* gene provides instructions for making one part (the alpha subunit) of an enzyme called 3-methylcrotonoyl-CoA carboxylase or 3-MCC. Alpha subunits join with smaller beta subunits made from the *MCCC2* gene; six of these pairings together form a functioning enzyme. The alpha subunit also includes a region for binding to the B vitamin biotin, which is required for the enzyme's function.

The 3-MCC enzyme is found in mitochondria, which are the energy-producing centers inside cells. This enzyme plays a critical role in breaking down proteins obtained from the diet. Specifically, it is responsible for the fourth step in the breakdown of leucine, an amino acid that is a building block of many proteins. This step converts a molecule called 3-methylcrotonyl-CoA to a molecule called 3-methylglutaconyl-CoA. Additional chemical reactions convert 3-methylglutaconyl-CoA into molecules that are later used for energy.

### Health Conditions Related to Genetic Changes

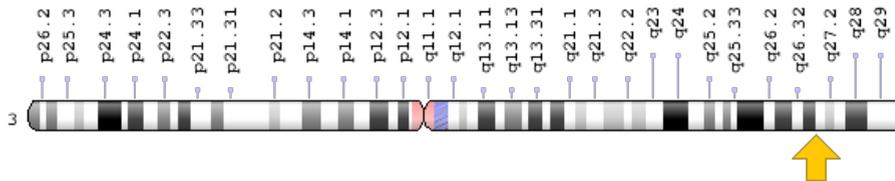
#### 3-methylcrotonyl-CoA carboxylase deficiency

At least 30 mutations in the *MCCC1* gene have been identified in people with 3-methylcrotonyl-CoA carboxylase deficiency (also known as 3-MCC deficiency). Most of these mutations change single amino acids in 3-MCC, but a few mutations lead to the production of an abnormally short version of the enzyme. Mutations in the *MCCC1* gene severely reduce or eliminate the activity of 3-MCC. As a result, leucine cannot be broken down properly, and byproducts of leucine processing build up to toxic levels in the body. These toxic substances can damage the brain, causing the characteristic signs and symptoms of 3-MCC deficiency.

## Chromosomal Location

Cytogenetic Location: 3q27.1, which is the long (q) arm of chromosome 3 at position 27.1

Molecular Location: base pairs 183,015,218 to 183,099,587 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- 3-methylcrotonyl-CoA carboxylase 1
- 3-methylcrotonyl-CoA carboxylase biotin-containing subunit
- MCCA
- MCCA\_HUMAN
- MCCase subunit alpha
- methylcrotonoyl-CoA carboxylase 1 (alpha)

## Additional Information & Resources

### Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Major pathways of branched-chain amino acid metabolism  
<https://www.ncbi.nlm.nih.gov/books/NBK20436/figure/A3097/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28MCCC1%5BTIAB%5D%29+OR+%28%28MCCA%5BTIAB%5D%29+OR+%283-methylcrotonyl-CoA+carboxylase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bm+h%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- 3-METHYLCROTONYL-CoA CARBOXYLASE 1  
<http://omim.org/entry/609010>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=MCCC1%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=6936](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6936)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/56922>
- UniProt  
<http://www.uniprot.org/uniprot/Q96RQ3>

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